



Mutational analysis of the RB1 gene in Moroccan patients with retinoblastoma

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Résumé en anglais	<p>PURPOSE: Retinoblastoma (RB), the most common intraocular tumor occurring in infancy and early childhood, is most often related to mutations in the RB1 gene. In this study, we screened the RB1 germline mutations in 41 unrelated Moroccan patients with retinoblastoma, 25 heritable cases, and 16 sporadic unilateral cases.</p> <p>METHODS: After complete ophthalmic examinations were performed and consent obtained, DNA was extracted from peripheral blood, and screening of RB1 mutations was performed with PCR direct sequencing of the promoter and the 27 coding exons of the RB1 gene.</p> <p>RESULTS: We identified ten germline mutations in 10/41 (24.39%) unrelated patients, among which three had not been previously reported. The mutation detection rate was 40% (10/25) in the heritable cases and 0% (0/16) in the sporadic unilateral cases. Of these mutations, six were nonsense, and three were frameshifts, all associated with severe phenotypes resulting in bilateral and multifocal tumors. One splice site mutation was found in a familial case associated with a low expressivity phenotype resulting in unilateral and unifocal tumors. Moreover, eight intronic variants were identified, three of which were novel.</p> <p>CONCLUSIONS: This first report of RB1 gene screening in Moroccan patients with retinoblastoma shows a comparable mutational spectrum to those reported previously, which has evident importance for managing patients with retinoblastoma and their families.</p>
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